SNiPAcard

Block annotations

Block info

genomic range	chr1:9,004,120-9,011,627
block size	7,508 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.432 [-1.57 – 4.333]	**gene(s) hit or close-by**	CA6
phastCons	μ = 0.180 [0 – 1]	**eQTL gene(s)**	–
GERP++	μ = 0.327 [-4.55 – 3.39]	**potentially regulated gene(s)**	CA6, RERE
CADD score	μ = 7.190 [0.168 – 23.5]	**disease gene(s)**	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CA6	missense variant	ENST00000377436	NM_001270500.1	ENSP00000366654	A/G	gCc/gGc	?	?	1
CA6	missense variant	ENST00000377443	NM_001215.3	ENSP00000366662	A/G	gCc/gGc	?	?	1
CA6	missense variant	ENST00000480186	?	ENSP00000435280	A/G	gCc/gGc	?	?	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000435401	1	ENST00000377436, ENST00000480186, ENST00000377443	CA6
FFCP00000435402	1	ENST00000377436, ENST00000480186, ENST00000377443	CA6

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000003284	1	ENCP00000000458	RERE

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000530170 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K27me3, DNase1
		blood (K562)	H3K27me3
		skin (NHEK)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CA6	upstream gene variant	1826	ENST00000377436	NM_001270500.1	ENSP00000366654	1
CA6	upstream gene variant	1806	ENST00000549778	?	ENSP00000447108	1
CA6	upstream gene variant	1822	ENST00000377443	NM_001215.3	ENSP00000366662	1
CA6	upstream gene variant	1821	ENST00000480186	?	ENSP00000435280	1
CA6	upstream gene variant	1807	ENST00000476083	?	?	1

CA6	upstream gene variant	1826	ENST00000377442	NM_001270501.1	ENSP00000366661	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
CA6	ENST00000377436	NM_001270500.1	ENSP00000366654	G	ggG/ggC	2
CA6	ENST00000480186	?	ENSP00000435280	2	2	2
CA6	ENST00000377443	NM_001215.3	ENSP00000366662	G	ggG/ggC	2

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CA6	ENST00000377436	NM_001270500.1	ENSP00000366654	4
CA6	ENST00000377442	NM_001270501.1	ENSP00000366661	5
CA6	ENST00000480186	?	ENSP00000435280	4
CA6	ENST00000377443	NM_001215.3	ENSP00000366662	4
CA6	ENST00000476083	?	?	5
CA6	ENST00000549778	?	ENSP00000447108	5